Journal of Genetic Counseling, Vol. 9, No. 5, 2000

Genetic Library

June Peters[1,2]

The themes of this library are multicultural counseling and research, public health, psychosocial issues in genetic counseling, and cancer genetics.

Bittner S, Bialek E, Nathiel S, Ringwald J, Tupper M (1999) An alternative to managed care: A "Guild" model for the independent practice of psychotherapy. *J Marit Fam Ther* 25:99–111.

Associations of independent, nonmanaged care psychotherapists are springing up in response to problems in practicing undermanaged care. Some of the problems experienced by therapists are common to genetic counseling, such as the demands by managed care for breaches in confidentiality, second-guessing assessment and treatment decisions that may not be in the best interest of the family, and concerns about conflicts of interest by payers. This paper describes the evolution and organization of one such guild whose members do not practice in the same office, are not mutually liable, and are not hierarchically structured by profession. They take a stand on commonly held values, such as privacy, choice, and tailoring the clinical encounter to meet the needs of the client, that will probably strike a chord with most genetic counselors. The clients pay a reduced fee out of pocket rather than billing the managed care organization. The idea of a multidisciplinary affiliation of clinicians who pool information and marketing resources could be one model for some genetic counselors in the next millennium.

Boyd J, Sonoda Y, Federici MG, Bogomolniy F, Rhei E, Maresco DL, et al. (2000). Clinicopathologic features of BRCA1-linked and sporadic ovarian cancer. *JAMA* 382:

[1]National Cancer Institute, Rockville, MD.
[2]Correspondence should be directed to June Peters, Clinical Genetics Branch, Division of Cancer Epidemiology and Genetics, NCI, 6120 Executive Blvd., Room EPS 7021, Rockville, MD 20852-7231; petersju@mail.nik.gov.

1059-7700/00/1000-0435$18.00/1

There is mounting evidence that some hereditary tumors may have a different biologic, histologic, cytogenetic, and molecular profile than sporadic cancers have and that these differences could lead to differences in the behavior of those tumors. This report supports the previous data about clinical behavior of tumors in both HNPCC-associated and BRCA-associated cancers. The authors conducted a retrospective cohort study of a consecutive series of 933 ovarian cancers diagnosed and treated at their comprehensive cancer center from 1986 to 1998. They focused on the 189 self-identified Jewish patients because of the ease of screening for the three common BRCA1/2 mutations in this population. They found 88 hereditary cases on the basis of molecular screening that identified one of the common mutations. The comparison group consisted of the 101 remaining subjects and patients from two clinical trials. Both groups may have also contained subjects with mutations that were not detected by their molecular screening method. This analysis demonstrated that histology, grade, stage, and success of cytoreductive surgery were similar for hereditary and sporadic cases. Few ovarian cancers were diagnosed prior to age 40 in any patient. However, the mean age at diagnosis was lower (54 vs. 62 years) for BRCA1 vs. BRCA2 patients. The hereditary group as compared to the controls had a longer disease-free interval following primary chemotherapy and those with advanced-stage disease had improved overall survival compared to control participants with advanced-stage disease. The authors' retrospective means of ascertainment avoided the selection bias of preferential inclusion of only living patients. Differences in outcome due to treatment were also reduced since all patients had been diagnosed and treated at the same institution over the same period. The authors speculate on possible treatment implications of the longer survival, due either to the tumors being more indolent and slow growing or to the possibility that BRCA-associated tumors may be more responsive to selected chemotherapy agents.

Dignam JJ, Colangelo L, Tian W, Jones J, Smith R, Wickerham DL, Wolmark N. Outcomes among African-Americans and Caucasians in colon cancer adjuvant therapy trials: Findings from the National Surgical Adjuvant Breast and Bowel Project. *J Natl Cancer Inst* 91:1933–1939.

Dignam et al. report the outcomes of adjuvant chemotherapy in five randomized clinical trials involving black patients with colon cancer compared with white patients with colon cancer. These studies confirm findings, first reported 30 years ago, that there are increasing disparities in cancer mortality between African-American and white Americans. In the US, race is a surrogate for socioeconomic class. When one considers that cancer-specific survival is equal when there is equal treatment, but overall survival is worse for African-Americans, we are left to conclude that there are different patterns of medical care for different segments of our population. Although medical research has found efficacious treatments

for some forms of cancer, society has not disseminated those treatments equally. Although we have less dramatic evidence, we know that these inequities hold true for genetic discoveries and services as well. The burden is on us to go beyond our traditional roles to help find and provide remedies for this unacceptable reality (See also Brawley OW, Freeman HP. (2000) Editorial: Race and outcomes: Is this the end of the beginning for minority health research? *Journal of the National Cancer Institute* 91:1908).

Gettig E, Baker T, Khoury MJ, Bryan, J, Pierce H, Puryear M, Thomson E. (1999) Report on the second national conference on genetics and public health: Genetics and disease prevention: Integrating genetics into public health policy, research, and practice in Baltimore, MD, Dec 6–8, 1999. *Commun Genet* 2:119–136.

This is a report of the proceedings of one of a series of recent conferences about the intersection of genetics and public health. Cosponsored by a variety of governmental agencies and private institutions, its aim was to increase public health professionals' understanding of the application of genetics to, and its effect on, public health activities. This was also an opportunity for on-going dialogue and planning for anticipated healthcare changes that may be elicited by advances in molecular genetics. The proceedings included both plenary sessions by scientific and public health luminaries as well as a number of workshops. Some of the important topics included developing public health capacity within a public health infrastructure, financing of public health genetic functions, and incorporating diverse community perspectives. This publication includes abstracts from presentations and workshops.

Pinsof WM, Wynne LC (2000) Toward progress research: Closing the gap between family therapy practice and research. *J Marit Fam Ther* 26:1–8.

The authors state in the opening sentence that "couple and family therapy research has had little, if any, impact on the practice of most couple and family therapists." The same might be said of some genetic counseling research. They cite a number of reasons and then present a more clinically relevant model for research that can change and inform practice. We might look at this experience to help shape goals for future genetic counseling research. The authors begin by making a differentiation between efficacy and effectiveness. Well-conducted efficacy research has all the characteristics (it occurs in a controlled setting, involves a treatment and control group, etc.) we have come to recognize in the clinical trial of a medical treatment. Some of the genetic counseling interventions for hereditary cancer susceptibility follow this model and are expected to yield informative results about what works under these strictly controlled conditions.

Genetic counseling certainly needs more of this type of efficacy research. The problems to this approach derive from the fact that it is so structured in its planning and application that it bears little resemblance to the actual practice of counseling. Therapeutic methods have become increasingly eclectic and integrative. Although informed by theory or a favored approach, the counselor's experience is organized in regard to individual and specific cases. For example, the session begins with the counselor approaching the client or family as a particular type of system, e.g., family with an autistic child. The counselor with knowledge from the literature of what works best, on average, for clients in this particular situation, starts off in a standardized way. However, subsequent direction of therapy is then guided less by the results of efficacy research and more by the clients' responses to the opening interventions. What comes next depends on what came before. Thus, good therapy most resembles disciplined improvisation.

The authors propose a broader study of effectiveness that assumes the possibility of alternative legitimate pathways for establishing the effectiveness of a treatment, e.g., including case study and qualitative methods, documenting narratives of change, and more longitudinal research. They give high priority to developing quantitative research methodologies that track change over the course of therapy. Their model of counseling is one of a collaborative learning process between counselor and client.

The authors call for a collaborative research program that oscillates between studying family change naturalistically and then moving into the laboratory to more formally test the hypotheses derived from those observations. They promote a concept of "progress research," which integrates process and outcome perspectives into a unified methodology that feeds research data back into the therapy where it can make a difference. It seems that this perspective offers opportunity for us to serve as the clinical arm of collaborative genetic counseling research of this type.

My criticisms of the paper are that the authors take a dominant culture view and they do not explicitly make an effort to address multiculturalism nor are they explicit about studying the person of the therapist as an important source of variability in the interventions. Nonetheless, the proposals that they have made could easily accommodate these issues.

Protheroe D, Turvey K, Horgan K, Benson E, Bowers D, House A. (1999) Stressful life events and difficulties and onset of breast cancer: Case control study. *Br J Med* 319:1027–1030.

The question of whether psychosocial factors influence the development of disease has been asked for centuries, especially regarding cancer. A number of factors have been studied, including "general liability to the buffets of ill fortune," personality, outward expression of character, mood disturbances especially

depression and anger, stress, coping, and adaptation; loss and bereavement; specific stressful life events; and prior long-standing difficulties. Results of these studies have been mixed and conflicting. The objective of this study was to determine the relation between stressful life events and difficulties and the onset of breast cancer. They used a case-control approach of studying women referred to a surgeon at 3 NHS breast clinics for evaluation of a breast lump. The cases were women found to have cancer, and the controls were women with normal or benign breast conditions. The authors calculated odds ratios of the risk of developing breast cancer after experiencing one or more severe life events, severe 2-year nonpersonal health difficulties, or severe personal health difficulty. The study found that women diagnosed with breast cancer were no more likely than the controls to have experienced any of these difficulties in the past five years. There are a number of methodological problems with this as with many other similar studies. The authors concluded that their findings do not support the hypothesis that severe life events or difficulties are associated with breast cancer diagnosis. This is in keeping with several other recent literature reviews and meta-analyses. If there is a role for stress in illness, it is not likely to be very straightforward.

Salovey P, Rothman, AJ, Detweiler JB, Steward WT. (2000) Emotional states and physical health. *Am Psychol* 55:110–121.

This paper takes up where the Taylor et al. paper (see later) left off in exploring potential mechanisms linking pleasant feelings and good health. The authors consider the evidence in a number of areas including (1) direct effects of positive affect on physiology, especially the immune system; (2) the information value of emotional experiences; (3) the psychological resources engendered by positive feeling states; (4) the ways in which mood can motivate health-relevant behaviors; and (5) the elicitation of social support. One section on emotional states and psychological resilience had material very relevant to genetic counseling. They state that "appropriate health practices are often difficult to carry out and psychologically taxing. In order to adopt precautions, people must first recognize the possibility that they are at risk for an unwanted health problem. Yet people actively strive to maintain their optimistic sense of their personal risk and resist acknowledging that they have health concerns. People who use illness screening or detection practices [e.g., prenatal diagnosis or annual mammography] must be willing to run the risk of learning that they have a health problem. Noncompliance with screening guidelines often reflects an unwillingness to face this risk. Positive emotional states can facilitate health behavioral practices by providing the resilience that people may need to confront the possibility that they might have or develop a serious health problem." Perhaps this speaks to an unarticulated goal of the genetic counselor's role in inspiring hope in others, particularly those struggling with present illness or the specter of future health problems.

Schrag D, Kuntz KM, Garber JE, Weeks JC (2000). Life expectancy gains from cancer prevention strategies for women with breast cancer and BRCA1 or BRCA1 mutations. *JAMA* 283:617–624.

This paper uses decision analysis to synthesize what is known about the efficacy of surgical and chemoprevention strategies. Looking at a hypothetical group of women, the authors' goal was to examine the survival effect of three methods of breast/ovarian cancer prevention: tamoxifen (tam), bilateral prophylactic oophorectomy (PO), prophylactic mastectomy (PM), or a combination of these. Seven strategies using combinations of tam, PO, and PM were compared with careful surveillance alone. The analyses were run for different penetrance figures for BRCA1 and BRCA2 mutations. The results indicate that life expectancy (LE) can be significantly increased for young women with early-stage breast cancer and moderately or highly penetrant BRCA1/2 mutations based on the prevention of second cancers by timely use of these strategies. More specifically, life expectancy is increased more for women who utilize the prevention options at younger ages than for older women. Gains in LE are greater for women with node-negative (i.e., early stage) disease. The life expectancy increments were much greater for women with BRCA mutations than for those with sporadic cancers. The authors note a few cautions. First, this study is based on modeling, which is highly dependent on the assumptions it incorporates. One such assumption is that tam is equally effective in women with BRCA mutations as in the clinical trial population. Another is that we know the penetrance of BRCA mutations. It behooves clinicians to keep updated on the most current information about mutation-specific cancer risks when considering how these estimates apply to individual patients. Also, survival is only one of the many factors that should enter into an important and irreversible decision such as prophylactic surgery. It is clear that more empirical data are needed on all of these issues. In the meantime, this paper provides another tool in the cancer genetic counselor's toolbag for facilitating medical decision-making in those with hereditary cancers.

Seligman MEP, Csikszentmihalyi M (2000) Positive psychology. *Am Psychol* 55:5–14.

This is the introductory article to an entire issue of the American Psychologist devoted to happiness, well-being, optimism, and other topics related to positive psychology. The authors have spent their careers bucking the system of clinical psychology that focuses primarily on pathology in favor of looking at the features that make life worth living, e.g., hope, wisdom, creativity, future-mindedness, courage, spirituality, responsibility, and perseverance. They consider these to be the human strengths that act as buffers against mental illness and bring us into the realms of work, education, growth, love, and play. The other papers in this series follow along

three main topics: (1) What makes one moment "better" than the next? (2) Defining the positive personality; and (3) Recognizing that people and experiences are imbedded in social contexts of communities and institutions, which involve interlocking social relationships. The implications for genetic counseling await study.

Sue S (1999) Science, ethnicity and bias: Where have we gone wrong? *Am Psychol* 54:1070–1077.

This author attempts to answer these questions: Is psychological science biased against ethnic minority research? Should ethnic minority researchers turn to alternatives other than traditional experimental and empirical methods? Why is there a relative lack of high quality ethnic minority research? Some of the questions and their answers apply to genetic counseling research as well as to psychology. Sue observes that much research on ethnic minority groups is of uneven or low quality and quantity and is underfunded. He believes that part of the reason for this disparity is the overemphasis on reliability (reproducibility) and internal validity (related to causality and credibility) over external validity (related to generalizability and the meaning of the results for better understanding of real people in real life situations) in peer review of research proposals and publications. This bias is operationalized in reviewer preferences for methodologies with more emphasis on tightly controlled experimental designs assuming a linear causal relationship of independent and dependent variables over the generalization from research conducted on diverse target populations directly in their actual environment with the accompanying noise and extraneous variables. The author concludes that science itself is not biased against ethnic research, but that the criteria used to judge the suitability of research for publication or funding are selectively enforced to steer away from the study of culture, which is ultimately a loss to us all. Sue advocates several remedies in order to develop stronger research on ethnic minority groups: (1) acknowledge that there are many types of knowledge and many ways of knowing, and that science is only one way; (2) require all research studies to address external validity issues and explicitly specify the populations to which the results apply, while maintaining strong internal validity standards. In other words, demand that all research show relevance for different groups; (3) include different research approaches into research proposals, e.g., qualitative and ethnographic methods; and (4) when ethnic differences are found in research results, require researchers to explain what aspects of ethnicity are responsible for the differences.

Sue DW, Bingham RP, Porche-Burke L, Vasquez M. (1999) The diversification of psychology: A multicultural revolution. *Am Psychol* 54:1061–1069.

Psychologists are becoming increasingly aware of the need for promoting diversity in practice in order to offer good care and to promote social justice.

This paper summarized major issues that emerged from the American Psychological Association's National Multicultural Conference and Summit held in 1999 in California. The agenda was broad: dealing with some consequences of diversification of the US population; struggling with how to facilitate difficult dialogues on race, gender, and sexual orientation; acknowledging the interconnectedness of spirituality and mental health; and recognizing that psychology operates within the paradigms of the dominant Eurocentric U.S. culture. Not surprisingly, the group concluded that an overhaul of training programs and educational models are needed. Many of the challenges to expanding multicultural education in psychology seem similar to those faced by genetic counseling training and continuing education efforts. Curriculum reform to incorporate cross-cultural competence is certainly necessary but not sufficient to produce change. The participants advocated numerous initiatives including those to increase faculty and student multicultural competencies; promote an inclusive and positive institutional and professional climate; diversify application processes to training programs to include core values related to multiculturalism; and identify and commit resources to increase diversity within the profession and patient populations.

Taylor SE, Kemeny ME, Reed GM, Bower JE, Gruenewald TL (2000) Psychological resources, positive illusions, and health. *Am Psychol* 55:99–109.

Shelley Taylor and colleagues at UCLA have long been known for their studies of the relationship between health and psychological beliefs such as optimism, personal control, and a sense of meaning. One of their most startling findings in women with breast cancer and men with AIDS was that positive attitudes that were illusory, e.g., unrealistic in the face of objective medical evidence of the degree and progression of illness, were predictive of longer survival. These results were replicated in conditions that controlled for a wide variety of potential predictors of death. While the authors acknowledge that we do not fully understand the biopsychosocial pathways by which such protective effects occur, the evidence is mounting that such positive beliefs not only help people adapt to stressful events more successfully, but may actually protect health.